Adapted from: APBIO Skipper

GENETICS ENRICHMENT PROJECT

Due 3/13/19

Adopt-a-Chromosome PROJECT

The focus of this project will be on <u>The Human Genome Project</u> and information readily available on the World Wide Web. There are two aspects to this project - First you will need to go online and access information and then you will need to express your knowledge and understanding in a *colorful*, *visually interesting and informative way*.

Chromosome POSTER (100 points)

- 1. A class lottery will help you to pick a human chromosome to get started you may also be assigned a "p" or "q" arm of a chromosome. This will only apply when you choose a gene and disease/disorder. You will research the entire chromosome for the beginning of the project.
- 2. Utilize the following web-sites for the required information
 - <u>A.</u> START HERE: Science: *A gene map of the human chromosomes*: or just use google images & find a visual representation of your assigned chromosome

http://web.ornl.gov/sci/techresources/Human Genome/education/index.shtml

http://vega.sanger.ac.uk/Homo sapiens/index.html

http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

http://www.ornl.gov/sci/techresources/Human Genome/posters/chromosome/chooser.shtml

- 3. Research the following aspects about your chromosome:
 - A. Basic information on your chromosome
 - 1. Size in base pairs/physical characteristics (centromere, banding)
 - 2. # of genes assigned to this chromosome
 - 3. Notable genes on a chromosome map give examples!! (if you're assigned an "arm" focus on your side only), give me loci, protein produced/characteristic or trait associated with & show location on your chromosome map highlight it with a highlighter on the map and write the description next to it on the poster
 - B. Pick ONE genetic disease that has been sequenced on your chromosome to study in detail (it's easiest to pick a gene that is associated with one disease/disorder, and that you are mildly interested in this disease/disorder):

Hint: pick a disease/syndrome/disorder that is only associated with a mutation in that gene

- 1. Research the disease using pubmed or pubmed health to find the gene associated with the disease you chose to focus on
- 2. Find the nucleotide sequence Download a copy!! **DO NOT PRINT ENTIRE SEQUENCE** unless it's short enough to place on your visual aide.
 - a. http://www.ncbi.nlm.nih.gov/sites/entrez
 - i. In the search menu select gene in the drop down menu and perform for you specific gene
 - ii. The top query result will most likely be your gene of interest, but check to make sure it is from *Homo sapiens*.
 - b. click on the gene and use FASTA or GENBANK to find the nucleotide sequence and its size - you can also get location on the chromosome (loci) and # of exons on the first page, before going to GENBANK or FASTA
- 3. What is the ~size of this gene (# of BP) /what is known about the physical structure of this gene (#Introns/ #Exons)? You may draw a map for this part (OR print one out) a banding pattern map is great here to designate locus
- 4. What is the size of the protein that it translates to (# of AA)? Download & print a copy, be sure to label start and stop codons and (if you want) the introns and exons if they aren't labeled already.
 - a. use ncbi again change dropdown menu to protein
 - b. use the name of the protein to search not the gene you can get this information when you search the gene (usually at the top of the gene results they have the protein name)

- c. find the amino acid sequence & print!
- 5. What kind of protein does this gene produce?? What is its function? What is its shape? What does it do? google is great for this or google image to find the crystallography image of your protein
- 6. Who are the scientists who worked on this disease? Where did they do their research? In what year(s) was this research done? Find an article.
- 7. A short summary of your disease in 1 paragraph or bullet point form. Pictures are helpful! Don't make me read a small paper I want the highlights
- 8. The mapped location of the gene on the chromosome. Notate its "loci" on your Chromosome map, or another map that is similar.
- 9. The protein sequence of the wild type (normal gene already done above) and the diseased version (mutation). Find the mutation that normally occurs (either in the nucleotide sequence or in protein production at the amino acid level) and explain or show in visual. DO NOT PRINT ENTIRE NUCLEOTIDE SEQUENCE!! You can focus only on the region or regions that mutation. What kind of mutation is it (inversion, deletion, substitution, repeat, etc.)
- 10. Visuals to help your reader understand the information you are providing; i.e. pictures of your disease, a picture of your folded protein in its active form, etc.

Be careful.

- · Warning! Warning! Choosing the disease is a crucial factor. Some will be easy and some very hard. You may want to change a few times depending on the availability of information. Pick a few genes on your designated chromosome to research, then choose the one with the most information available.
- For example, if you wanted to research deafness on chromosome 22, you would type in "deafness". Since many chromosomes carry genes that may result in deafness, you will find that many choices pop up. Make sure you find the right gene on chromosome 22. Look in the position column. 22p16 means that this gene is on chromosome 22 on the upper arm (p) a certain distance from the centromere. (q) means it is on the lower arm.
- As stated the OMIM number will provide you with lots of information. One bit of info is the "gene map locus" address of your chosen genes.
- Select the choice and click on the blue NM number. (for example, NM 022965) The resulting page will give you the DNA that codes for the mRNA (no introns) and the proteins. Below is a list of the amino acid abbreviations to help you. NCBI can be hard to maneuver. Ask Mrs. Hale! Use the drop-down menus at the top of ncbi.gov to narrow down your searches, i.e. pick gene when you're researching the gene and protein when your search word or phrase is the name of the protein, not the gene.

Amino Acid Abbreviations for NCBI printouts

Alanine A	Leucine L		
Arginine R	Lysine K		
Asparagine N	Methionine M		
Aspartic acid D	Phenylalanine F		
Cysteine C	Proline P		
Glutamic acid E	Serine S		
Glutamine Q	Threonine T		
Glycine G	Tryptophan W		
Histidine H	Tyrosine Y		
Isoleucine I	Valine V		

CREATE a POSTER FOR your CHROMOSOME ON STANDARD SIZE POSTERBOARD. These will be displayed.

Include as much information as you FEEL IMPORTANT. creativity counts!!!

Turn in your poster & bibliography (on the back of poster)

Topic:			
	Rating	X	= Points
Basic Information - (80 Pts)			
RATING SCALE:		1	
5 = Excellent			
4 = Good			
3 = Average			
2 = Poor			
1 = Unacceptable			
0 = Not Present			
Title Clear, organized & visually interesting			
Basic information of your chromosome.		4	
Map, other genes of interest, size, etc			
Visual map of chromosome.		3	
Picture and explanation			
Nucleotide sequence of gene of interest.		2	
With mutation information, exons/introns, bp #			
Basic information on gene of interest.		4	
What is the protein, what does it do, disease info			
Amino acid sequence of gene of interest.		1	
# AAs & visual of sequence			
TOTAL POINTS			
Poster Appearance (20 Points)			
Poster is colorful and neat with correct grammar		3	
and spelling, creative, accurate & eye-catching.			
Bibliography		1	
TOTAL POINTS			